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Sponastrime Dysplasia: Five New Cases and Review of Nine Previously Published Cases

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Sponastrime dysplasia (SD) is a dwarfing autosomal recessive short-limb bone dysplasia. The diagnosis is established by a combination of clinical and radiological findings of which the radiological are the more specific. The current diagnostic criteria are ambiguous as demonstrated by the fact that, in our opinion, three of the five patients reported since the original article do not have this condition. Comparison of our five patients and the 9 published patients has led to development of more specific diagnostic criteria. Previously undescribed complications of this condition are subglottic stenosis and tracheo-broncho-malacia, developmental coxa vara, and avascular necrosis of the capital femoral epiphyses.

KEY WORDS: dwarfism, autosomal recessive, developmental coxa vara, familial, osteochondrodysplasia

INTRODUCTION

Sponastrime dysplasia (SD) is a mild to moderately dwarfing bone dysplasia originally described and named by Fanconi et al. in 1983. The term sponastrime is an acronym derived from *spo*ndylar and *na*sal alterations with *stri*ations of the *me*taphyses. Nine patients have been published as examples of this condition. We believe that three of them do not have SD. On the basis of comparison of our five patients with those previously published, it appears that a unique combination of clinical and radiological findings is diagnostic from early childhood through mid-adolescence and examination of one newborn infant suggests unique combination of findings in the vertebral bodies and proximal femora at this age.

SUBJECTS AND METHODS

Nine patients have been published with a diagnosis of sponastrime dysplasia. We report five additional patients. Clinical and radiographic data from birth to 18 years of age on these 14 patients were reviewed to establish the clinical and radiographic characteristics of this dysplasia.

CASE REPORTS

Kindred I

Case 1. A.L. presented to the Oregon Health Sciences University at age 10½ months for evaluation of short stature. She was the product of a term pregnancy with a birth weight of 4.53 kg and a birth length of 48 cm. Evaluation demonstrated short stature (5th centile height) (arm span 66 cm, height 62.4 cm, lower segment 26.5 cm, and upper segment/lower segment ratio 1.36) (normal = 1.50). Results of chemistry battery, thyroid studies, and urinalysis were normal. Chromosomes were normal. The bone age at age 10.5 months was 8 months. Bone age was 18 months at 26 months of age. Studies were compatible with growth hormone deficiency on the basis of an insulin-arginine growth hormone stimulation test. Her peak growth hormone level was 6.0 ng/ml, diagnostic of biochemical growth hormone deficiency. She was treated with growth hormone beginning at 27 months continuing to the present at age 8. Her growth velocity is normal, but there has been no catch-up growth.

Examination at 6 8/12 years demonstrated short-limb short stature (height 102 cm and span 109 cm upper/lower segment ratio = 1.30). Intelligence was normal. Her facial appearance was characterized by mid-face depression, saddle-shaped nose, and epicanthal folds. She had increased lordosis and generalized joint laxity except for slight flexion contracture of the elbows. She had mild symptomatic bowing of the distal left tibia.

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Dedicated to Jürgen W. Spranger on the occasion of his 65th birthday with admiration and best wishes.

There was no family history of short stature or bone dysplasia. The father is 182.9 cm and the mother is 177.8 cm tall. There is a normal brother. A sister was subsequently born who was diagnosed as having SD at age 5 months (Case 2).

At age 7 a long leg brace was used to control a varus deformity below the knee. The brace was worn sporadically and discarded. The deformity had corrected by age 8½ years when last seen.

Radiographic findings will be described in the radiological findings section.

Case 2. A.L., sister of Case 1, was evaluated at age 5 months for short stature (under 3rd centile). Her birth weight was 2.8 kg and length 45.5 cm.

Examination demonstrated short-limb short stature, depressed mid-face, epicanthal folds, generalized joint laxity, and lumbar lordosis. Intelligence was normal. Her height was 53.5 cm, span was 54 cm, upper seg was 31.5 cm, and lower seg was 22.0 cm, upper/lower segment ratio = 1.45. Results of screening chemistry, growth hormone studies (peak = 30.7 ng/ml with levodopa stimulation), and a chromosome analysis were normal. Radiographic findings were similar, but less severe than those of her older sister and will be described in the radiological findings section. At age 2 years she developed a waddling gait and mild discomfort. Radiographs demonstrate right developmental coxa vara. The coxa vara had progressed moderately and had become bilateral when last evaluated at age 3 3/12 years (Fig. 1A).

Kindred II

Case 3. V.E. was evaluated at the duPont Institute, Wilmington, DE for short stature at age 2 years. He was the product of a normal pregnancy and delivery with a birth length of 44 cm and weight of 2.6 kg. He was evaluated in infancy for short-limb short stature and an unusual facial appearance. The evaluation included chromosome, urinary amino and organic acids, MPS screen, lysosomal enzymes, I-cell studies, WBC inclusions, and skin fibroblast culture, all being normal. At age 7 7/12 years his facial appearance was characterized by bifrontal bossing, mid-face hypoplasia, epicanthal folds, and mild prognathism. He had mild generalized joint laxity and genu valgum. Ophthalmological findings were normal. He demonstrated occasional premature ventricular contractions but had no clinical cardiac disease. His height was 100.7 cm, span was 109.3 cm, and US/LS was 53.4/46.3. Intelligence was normal. His parents' heights are 157 cm and 163 cm and an older brother is of normal height for age. Radiographic findings will be described in the radiological findings section. Mild hypothyroidism has been treated by synthroid since age 18 months with little effect on statural velocity.

Case 4. S.E. is a younger brother of V.E. He is the product of a normal pregnancy and delivery with a birth weight of 2.7 kg and length of 43 cm. Chromosomal findings were normal. Cryptorchidism was treated with HCG injection with testicular descent. Persistent pulmonary dysfunction led to a diagnosis of subglottic stenosis and tracheo-broncho-malacia. A limp at age 25 months prompted radiographs demonstrating progressive, unilateral coxa vara that was later treated by osteotomy.

When examined at age 5 9/12 years, measurements included height of 85.6 cm, span of 109.3 cm, US/LS of 54.4/32.2, and weight of 19.5 kg. He had bifrontal bossing, mid-face hypoplasia, prominent mandible, mild pectus excavatum, shortness of the left leg, mild genu valgum, 25° limitation of elbow extension, and mild generalized joint laxity. Intelligence was normal. Radiographic findings will be described in the radiological findings section (Fig. 1B).

Kindred III

Case 5. J.S. presented to the Children's Health Care-St. Paul, St. Paul, Minnesota at age 14.3 years, for evaluation of short stature. His birth weight was 2.9 kg. He had a fraternal twin sister with a birth weight of 3.1 kg. His mother was 159 cm and his father was 182 cm tall. He had a brother and sister of normal height.

On examination he was 138.4 cm tall (greater than −4 standard deviation). His span was 134.3 cm and the upper/lower segment ratio was 1.07 (normal for age is .97). He had a flat midface with a depressed nasal bridge and a somewhat prominent forehead. The nose was short and broad with a midline depression in the columella. His intelligence was normal. His genitalia were prepubertal.

Laboratory studies included a bone age of 14 years at C.A. 14 3/12 years, T4 of 8.1 ng/ml, and T3 uptake of 31.7%. Prolactin was 3 ng/ml and antigliadin antibody was negative. The peak growth hormone level after 20 minutes of exercise was 26 ng/ml (normal >10.0). An MRI of the head was normal. The LHRH stimulation test was normal. He was treated with depotestosterone 100 mg IM every 4 weeks for 12 doses with a resultant height increase of 8.3 cm in 12 months. He was treated with synthetic growth hormone at age 15 years 11 months (bone age 15 6/12 years) and was poorly compliant with a resultant height increase of 2.1 cm in 8 months.

At age 17 7/12 years, his height was 154.0 cm (greater than −4 standard deviation), weight was 56.59 kg, pubertal development Tanner V and his testes were 15 cc bilaterally. Radiographic findings will be described in the radiological findings section.

Prior Reports of Sponastrime Dysplasia

In 1983, Fanconi et al. described SD as a new type of skeletal dysplasia. They reported four sibs with dwarfism, normal intelligence, vertebral abnormalities, striated metaphyses, increased lordosis, frontal bossing, saddle nose, and a relatively large head.

Lachman et al. described two sibs with SD in 1989. Their patients had short stature, normal intelligence, vertebral abnormality, lumbar lordosis, striated metaphyses, mid-face hypoplasia, saddle nose, and prominent forehead. Each of their patients had been treated by a Harrington rod for thoracic kyphoscoliosis and demonstrated restrictions of elbow rotation. Iliac crest biopsy demonstrated abnormality in cartilage formation.

Camera et al. [1993] described two sisters with short stature and mid-face hypoplasia, metaphyseal striation,

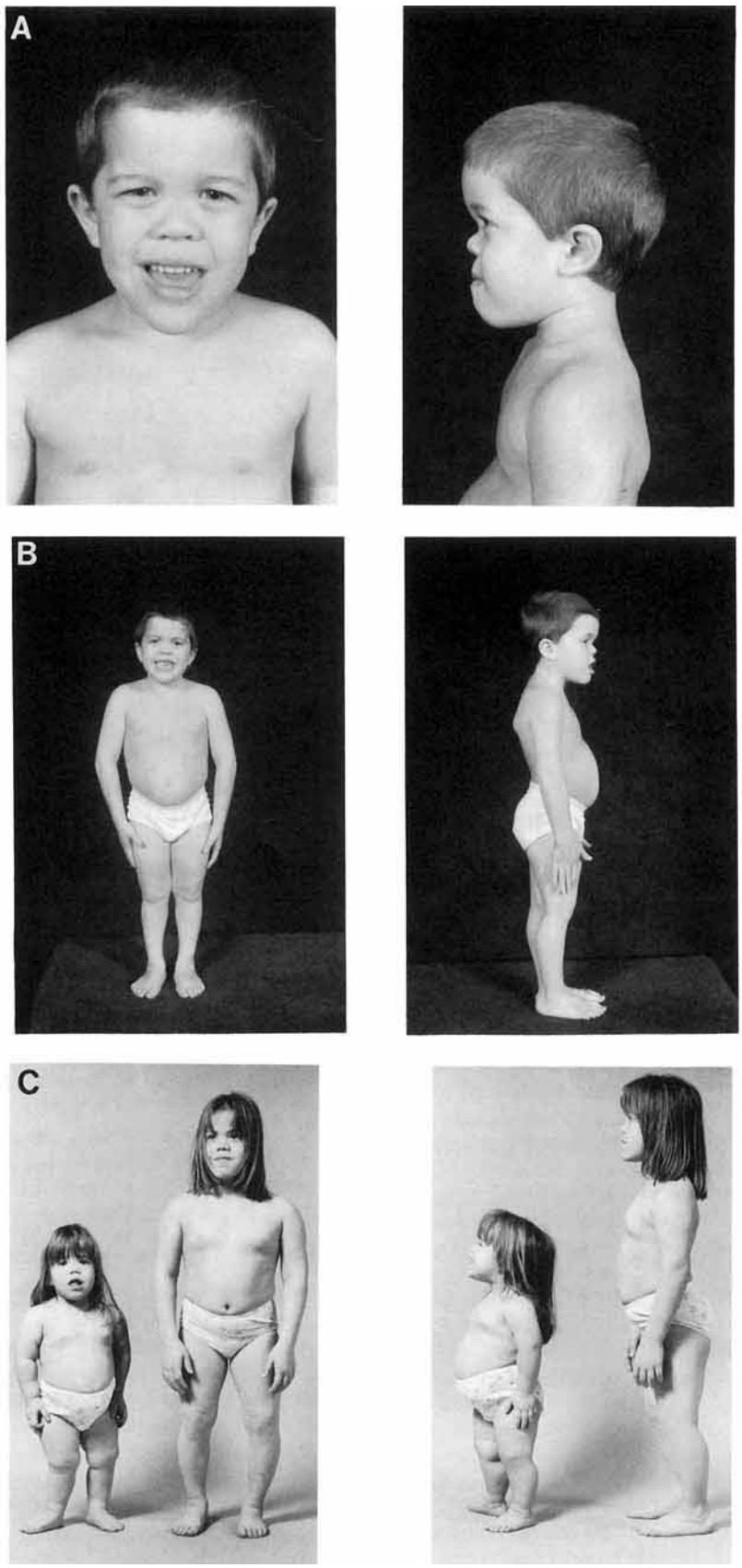

Fig. 1. **A:** AP and lateral photograph of patient 4 demonstrates mid-face hypoplasia, prominent forehead, and mild prognathism. **B:** AP and lateral standing photograph of patient 3 demonstrates short-limb short stature, and increased lordosis. **C:** Photograph of patients 1 and 2 demonstrates mid-face hypoplasia and angular deformity of the legs.

severe mental retardation, and vertebral abnormalities whom they felt had SD. Camera et al. [1994] also reported a boy with short stature, saddle nose, mid-face hypoplasia, absent patellar ossification, striated metaphyses, delayed carpal ossifications, and vertebral abnormality. We believe the patients reported by Camera do not have the radiographic characteristics of SD.

Clinical Features of Sponastrime Dysplasia

Growth. The patients of Fanconi et al. [1983], Lachman et al. [1989], and our patients all had mild to moderate dwarfing and almost all are short limbed. The lower limbs are more involved than the upper. Birth length is reduced but may be in the normal range. Growth failure is apparent early in infancy and is progressive. The tallest affected adult male was 154 cm and the tallest female 145 cm.

Facial appearance. All patients have a depressed nasal bridge and mid-face hypoplasia, epicanthal folds, prominent forehead, and most have a short broad anteverted nose, and mild prognathism. However, these traits are not unique to SD.

Other Clinical Findings

All of the patients that we believe have SD are of normal intelligence. Increased lumbar lordosis and generalized joint laxity were present in most patients. Several patients had limitation in elbow extension or rotation. Mild angular deformity of the lower limbs is common. Scoliosis requiring surgical treatment was present in two sibs.

Two patients with progressive infantile coxa vara have or will require surgical correction. Avascular necrosis of the hip was described in two young patients. Subglottic stenosis and bronchotracheomalacia occurred in one patient and biochemical hGH deficiency which did not have a typical response to treatment was found in one patient. In two sibs chemical hypothyroidism was treated but resulted in no appreciable changes in statural growth velocity.

Clinical manifestations may be highly suggestive of SD, but the diagnosis can only be made if characteristic radiographic changes are also present.

Radiological Findings

Changes in the lumbar vertebral bodies as seen in lateral projection are the most consistent findings in SD. The shape of the lumbar vertebral bodies is abnormal from birth through mid-adolescence with a predictable evolution over time. At all ages there is some variability in the shape of the bodies. The most common shapes at each age are described. Metaphyseal abnormalities are not as consistent as the lumbar body changes and may be absent. The distal femur and proximal tibia are the sites of major involvement. The distal tibia and fibula, the distal radius and ulna, and the proximal humerus and femur may also be involved and in that order of frequency.

Findings in the Lumbar Vertebral Bodies at Different Ages

Birth to approximately 6 months. The height of the vertebral bodies is markedly reduced. The anterior part of the body is taller than the posterior part and has convex endplates and a central anterior bony protuberance. The posterior part has straight endplates (Fig. 2A).

Approximately 6–24 months. The bodies are relatively taller than in the newborn infant. The anterior part is taller than the posterior part and has convex endplates. The posterior part has straight or slightly convex endplates. There is a distinct junction between the convex anterior endplates and the essentially straight posterior endplates. The posterior surface of the body is concave (Fig. 2B).

Approximately 15–20 months to 6 years. The anterior part of the body has convex endplates and is usually taller than the posterior part. The posterior part

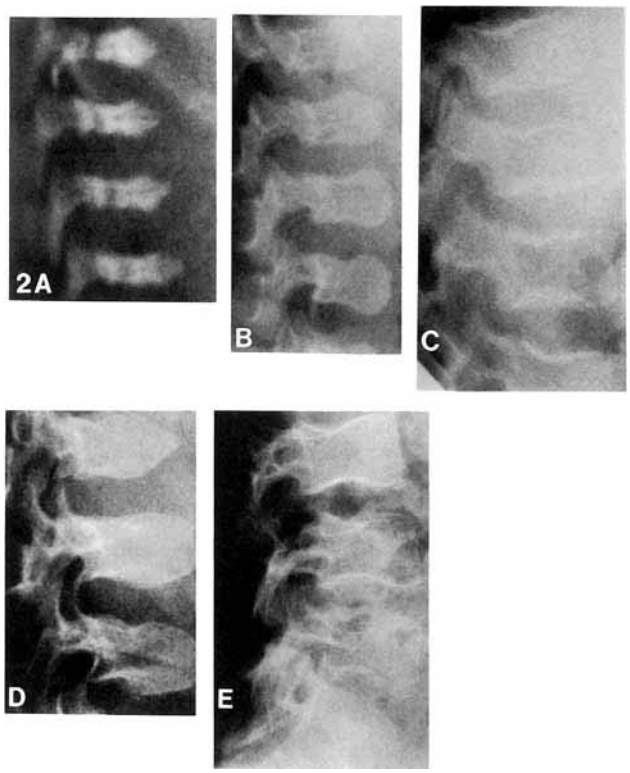

Fig. 2. **A:** Patient 4 at birth. Reduced height of vertebral bodies. The anterior part of the body is taller than the posterior part and has convex endplates and a central anterior bony protuberance. The posterior part has straight endplates. **B:** Patient 3 at 13 months. Bodies relatively taller. The anterior part is taller than the posterior and has convex endplates. The posterior part has straight or slightly convex endplates. There is a distinct junction between the anterior and posterior parts. The posterior surface of the body is concave. **C:** Patient 2 at 36 months. The anterior part is usually taller and has convex endplates. The posterior part has slightly convex endplates that tilt so the junction of the anterior and posterior part is the narrowest part of the body. The posterior surface of the body is concave. See text regarding the second common shape. **D:** Patient 1 at 7 years. The relative height of the body increases with age. The posterior-most part may be as tall as the anterior part. The posterior surface of the body is concave. The endplates show a smooth concavity in the posterior two thirds to three fourths of the body. The endplates anterior to this are convex. The center of the endplate concavity is usually posterior to the midline of the body. **E:** Patient 5 at 15½ years. The bodies are usually of normal height and tend to have a uniform shape. The appearance of endplate concavities is unchanged from mid-late childhood (D, above). The posterior surface of the bodies is straight and the disk spaces are narrower than at an earlier age.

has slightly convex endplates that tilt so that the junction of the anterior and posterior parts is the narrowest part of the body. The posterior surface of the body is concave (Fig. 2C). Another pattern, the posteriorly wedged body shape, can also be seen at this age. The anterior part of the body is taller and has convex endplates. The posterior part of the body becomes progressively shorter from front to back. The posterior endplates are essentially straight and there is no clear-cut junction between the anterior and posterior endplates. A typical example is not seen in our material (see Fanconi et al. [1983] and Lachman et al. [1989] for illustrations of 2½ year old patients).

Approximately 4–6 years to 10–12 years. The relative height of the bodies increases with age. The posterior most part may be as tall as the anterior part. The posterior surface of the body is concave. The endplates show a smooth concavity in the posterior two thirds to three fourths of the body. The endplates anterior to this are convex. The body's narrowest point (the center of the endplate concavity) is usually posterior to the midline of the body (Fig. 2D).

Approximately 10–12 to 15–16 years. There is a normal height-to-length relationship in the vertebral bodies. The appearance of the concavity of the endplates is unchanged from that seen in mid-to-late childhood. The posterior surface of the body is straight. The disk space is narrower than at an earlier age (Fig. 2E).

There is no lateral lumbar spine radiograph of a skeletally mature individual, and there is variability in the endplate deformity in the 15 5/12- and 18 3/12-year-old patients of Fanconi et al. [1983]. Thus, the appearance of the lumbar bodies in the adult is unknown.

Findings in the Metaphyses at Different Ages

These abnormalities consist of irregularity of the metaphyseal margin, density either band-like or localized adjacent to the metaphyseal margin, and vertically oriented striations that run between the metaphyseal margin or the adjacent density into the metaphysis and may extend into the diaphysis. There was variability in magnitude of all of these changes in the two sets of sibs whom we followed serially. Patients 1 and 2 had greater metaphyseal margin abnormalities than patients 3 and 4 and ultimately had greater vertically oriented abnormalities away from the metaphyseal margin and extending into the diaphysis. Some of these changes were not striations in the usual sense of the word but undoubtedly had a similar pathogenesis.

An exception to the generalizations above were the abnormalities in the newborn and young infant. The changes at this age consisted of an unusual shape for the proximal femora. There is a prominent bony protrusion in the region of the lesser trochanter and absence of the normal metaphyseal flare at the upper end of the femur as seen in AP projection. In oblique projection the bony protrusion is prominent and the femur proximal to it is somewhat lucent.

Lateral projection shows that the proximal femur is beveled in the sagittal plane and is narrowest at its proximal end. The configuration is similar to that present in achondroplasia, but less marked (Fig. 3A,B). No other significant bony abnormality was noted on skeletal survey. The deformity of the femur disappears by 18–24 months.

Around this time the distal femoral metaphyseal margins may be minimally irregular. There is often a dense band adjacent to all or part of the metaphyseal margin (Figs. 3C, 4A). This can be seen in normal individuals but is not common. Most young children with SD had this finding. Thereafter, there is an increase in the irregularity of the metaphyseal margin and the band-like density is replaced by a more patchy mixed density and lucency adjacent to the metaphyseal margin. At the time vertical longitudinal striations in the metaphyseal region become visible, they look like and probably are prominent compression trabeculae, a finding seen in osteopenic individuals and in some apparently normal individuals (Fig. 4B,C). Usually around 5 or 6 years there are abnormal metaphyseal margin changes and striations (Fig. 4D,E). The striations may extend beyond the metaphysis into the adjacent diaphysis. In our patients these changes become more marked with increasing age. Our oldest patient was studied at 8 3/12 years (Fig. 4F). In one patient of Lachman et al. [1989], there was density of the metaphyseal margins and striations at age 5½ years. Their other patient had a broad horizontal density adjacent to the metaphyseal margin at age 3 years and no metaphyseal abnormality at age 9 years.

Other radiological changes included absence of the normal childhood increase in the interpediculate distance from L1 to L5. This abnormality was present in studies at ages 2 years and older. It could not be evalu-

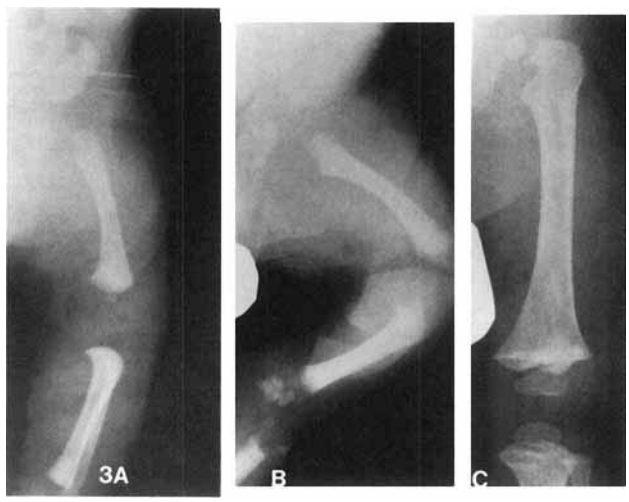

Fig. 3. **A,B:** Patient 4 at birth. A: Deformity of proximal femur in A-P projection with bony protrusion in region of lesser trochanter and lack of normal metaphyseal flaring at proximal end of femur. B: Appearance in oblique projection. Deformity is similar, but less marked than that in proximal femur in achondroplasia. **C:** Patient 4 at age 24 months. Slight irregularity of distal metaphyseal margin with density adjacent to it. Note deformity of proximal femur due to developmental coxa vara.

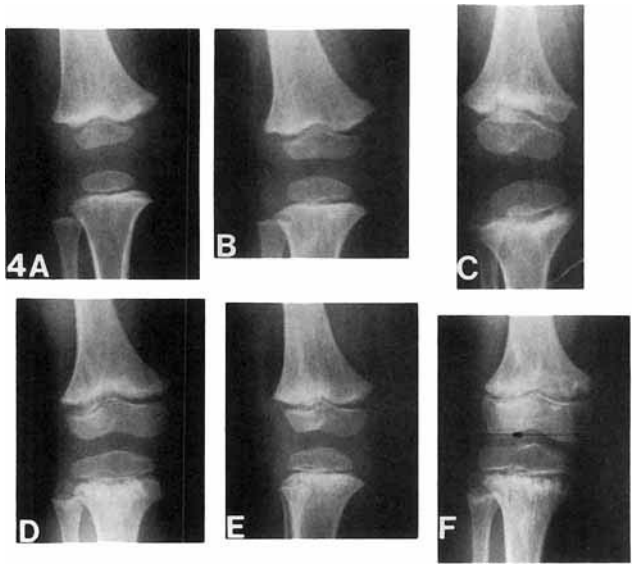

Fig. 4. **A:** Patient 3 at age 24 months. Dense areas adjacent to medial part of distal femoral metaphyseal margin which is minimally irregular. **B:** Patient 3 at age 3½ years. Minimal metaphyseal margin irregularity continues. The density adjacent to the margin is no longer present. **C:** Patient 2 at age 36 months. Marked irregularity of the femoral and tibial metaphyseal margins with some adjacent areas of density. Vertical longitudinal densities (striations) are indistinguishable from compression trabeculae. **D:** Patient 1 at 5 years. Marked irregularity of femoral and tibial metaphyseal margins with adjacent densities. The tibia is more involved than the femur. There are one or two abnormal striations in the lateral aspect of the femur. **E:** Patient 3 at age 6½ years. There is slightly more irregularity of the femoral metaphyseal margin than was present at age 3 years. Minimal juxtamarginal density is present. A few questionably abnormal striations are present in the femur. **F:** Patient 1 at age 8 3/12 years. Greater irregularity of the metaphyseal regions with smaller, but more sharply defined juxtamarginal areas of density compared to the appearance at age 5 years. There are now definitely abnormal striations and other vertically oriented densities in the metaphyses, some of which extend into the diaphysis.

ated at a younger age in our patients. Retardation of the carpal bone ossification was present in childhood. This is a finding in a number of bone dysplasias. It may result in small bones in the proximal carpal row in adulthood. Lateral skull radiographs showed a flattened maxilla and nasal bridge and relative prognathism with protrusion of the anterior teeth. As noted in the clinical section, there may be scoliosis which can be progressive. There was developmental coxa vara in two patients and avascular necrosis in the capital femoral epiphyses in two patients.

Genetics

Ten of the eleven patients with the diagnostic criteria for SD were sibs. All of the parents were clinically normal; none were consanguineous. Sponastrime dysplasia appears to be an autosomal recessive trait.

DISCUSSION

We evaluated serial radiographic examinations in two sibs in each of two unrelated families and a single patient in a third family. The two sets of sibs were examined periodically between the day of birth and age 8½ years. The single patient was 15½ and 17 years old when examined.

There are nine published cases. The patients of Fanconi et al. [1983] were four sisters examined once at ages 2 5/12, 13 3/12, 15 5/12, and 18 3/12 years, respectively. The patients of Lachman et al. [1989] were sibs who were examined several times between the ages of 2 9/12 years and 11½ years. They showed the same radiographic and clinical findings seen in the sisters of Fanconi et al. and in our patients. On the basis of these patients, Langer et al. [1996] suggested diagnostic criteria in which the age related radiological lumbar vertebral body changes were the most specific criterion.

Camera et al. reported two sisters [1993] and a male [1994] as examples of SD. We do not believe that these three patients have SD, primarily because they did not have the same radiological findings as the patients of Fanconi et al. [1983], Lachman et al. [1989], and our patients.

Fanconi et al. [1983] were impressed by the vertical striations in the adolescent sisters and referred to the striations in the name they gave the condition. We found that striations were usually not present in early childhood and that if they were present, they were not prominent and could not be confidently differentiated from compression trabeculae for several more years. Abnormal striations were present in late childhood. In most patients the more impressive abnormality was in the metaphyseal margin. However, our oldest patient with striations was age 8 3/12 years. We had no patients old enough so that physeal closure was imminent or had occurred, but we assume that the density and irregularity at the metaphyseal margin will disappear close to the time of physeal closure. The illustrations of the patients' knee radiographs of Fanconi et al. [1983] show striations and changes in the metaphyseal margins in the 13 year old and only striations in the 18 year old. Two patients, one patient of Lachman et al. [1989] and our 15-year-old patient, had normal appearing metaphyses.

In 1993, Camera et al. [1993] reported two adolescent sisters as examples of SD. They had frontal bossing and saddle noses similar to SD. The condition they had is most likely an autosomal recessive trait. They differed clinically in that they were severely mentally retarded in contrast to the patients of Fanconi et al. [1983], Lachman et al. [1989], and our patients, who had normal intelligence. There was a significant difference in height. The older patient of Camera et al. was 118 cm tall close to the time of skeletal maturity. The oldest patient of Fanconi et al. [1983], also a woman close to skeletal maturity, was approximately 145 cm tall.

Our principal reason for concluding that the patients of Camera et al. did not have SD is the difference in radiological findings. One sister was radiographed at 7 and 14 years and the other at 5 and 12 years. At both the younger and older ages the only abnormalities they had at the knee were fine vertical metaphyseal striations while the patient of Lachman et al. [1989] and our patients at the younger ages had abnormalities of the metaphyseal margins as well as striations. The age re-

lated abnormalities of the lumbar vertebral bodies were the most constant radiological findings in the patients of Fanconi et al. [1983], Lachman et al. [1989], and our patients. The lumbar bodies of Camera et al. did not show these changes at either the younger or older ages.

Verloes et al. [1995] reported a severely mentally retarded 10-year-old boy whom they thought might have the same condition as the sisters of Camera et al. Their patient had severe osteopenia and, as is common in this situation, the illustrations show prominent trabeculae in various normal orientations in the elbow region. There are vertical trabeculae in the epiphyses but not in the metaphyses at the knees. Their patient did not have the vertebral body abnormalities seen in the patients of Fanconi et al. [1983], Lachman et al. [1989], and our patients.

Verloes et al. [1995] felt that their patient and the sisters of Camera et al. did not have the same condition as the patients of Fanconi et al. [1983] and Lachman et al. [1989], and suggested that their patient's condition be called sponastrime dysplasia, type II. We do not think that this name is optimal. The three individuals suggested as having SD, type II do not have the pattern of radiological abnormalities demonstrated in the 11 patients who do look alike [Fanconi et al., 1983; Lachman et al., 1989 and our patients]. We think that the proposed terminology is more likely to further confuse rather than clarify the situation.

The boy reported by Camera et al. [1994] as an example of SD has the radiological findings of an incompletely delineated bone dysplasia that we have seen no reference to in the medical literature. The clinical findings in this condition are similar to those of SD. One of us (LOL) has seen studies on six patients with this condition including studies in childhood and adulthood in two patients. The illustrations of the radiographs of the knees, hands, and hips in the article of Camera et al. [1994] are diagnostic of this condition (Fig. 5A,B,C). The patients of Camera et al. did not have the vertebral body changes of SD.

We know of no other bone dysplasia that from late infancy to mid-adolescence has the combination of mild to moderate dwarfism, the typical facial appearance, and the sequence of age-related lumbar vertebral body changes seen in SD. An individual with an undiagnosed metaphyseal chondrodysplasia should be evaluated for SD by obtaining a lateral radiograph of the lumbar spine. At birth and in early infancy the suggestion is that diagnostic findings are present, but only one patient has been studied.

Conceivably, in later childhood and adolescence, sickle cell anemia or conditions causing osteomalacia or osteoporosis could result in lumbar bodies having the same shape seen in SD. It should be possible to distinguish these conditions from SD by characteristic clinical, laboratory, and imaging findings and by the absence of the clinical findings of SD.

Characteristic metaphyseal changes confirm the diagnosis. However, several affected individuals had no metaphyseal abnormalities, and their presence is not necessary to establish the diagnosis.

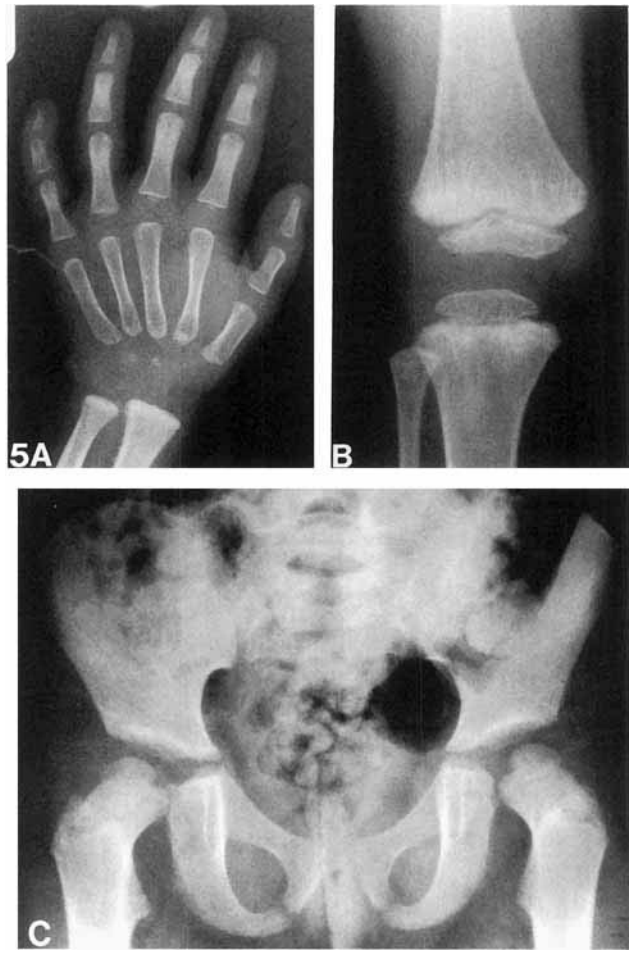

Fig. 5. **A:** Three-and-a-half-year-old girl. Narrow normally modeled metacarpals and retarded ossification of the epiphyses and carpal bones. **B:** Four-year-old girl. Abnormality of the metaphyseal margin and striations and small epiphyses in the knee region. **C:** Four-year-old girl. Normal pelvic bones. Small capital femoral epiphyses. Compare these illustrations with those in Camera et al. [1994].

We do not think that sponastrime dysplasia is a very useful term for this bone dysplasia. It suggests nothing to someone who is not familiar with it. If you are aware of the meaning of this term, it is somewhat misleading in that the only specific abnormality referred to in this acronym is the metaphyseal striations. These are not present in infancy or early childhood, and later in childhood they may be the least conspicuous metaphyseal abnormality. Two of the eleven patients did not have striations on examinations at an older age.

We think that it would be more helpful to call the condition a spondylometaphyseal dysplasia. The advantage of this kind of nomenclature is that someone who does not recognize the condition can see what regions of the osseous system show the greatest radiological abnormality and then can look at the conditions described under these anatomic regions in an atlas, textbook, or computer program. We suggest the name *spondylo*metaphyseal dysplasia, hypoplastic nasal bridge and midface type. Italicizing the *spondylo* in the name indicates that the spine changes are diagnostically more

important than the metaphyseal changes which may be absent in this condition.

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